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SNP alleles in human disease and evolution.

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In two randomly selected human genomes, 99.9% of the DNA sequence is identical. The remaining 0.1% of DNA contains sequence variations. The most common type of such variation is called a single-nucleotide polymorphism, or SNP. SNPs are highly abundant, stable, and distributed throughout the genome. These variations are associated with diversity in the population, individuality, susceptibility to diseases, and individual response to medicine. Recently, it has been suggested that SNPs can be used for homogeneity testing and pharmacogenetic studies and to identify and map complex, common diseases such as high blood pressure, diabetes, and heart disease. Consistent with this proposal is the identification of the patterns of SNPs in conditions such as diabetes, schizophrenia, and blood-pressure homeostasis. Although these studies have provided insight into the nature of human sequence variation, it is not known at present whether these variations are truly significant toxicologically and pharmacologically. Moreover, it is possible that most complex, common disorders are caused by the combined effects of multigenes and nongenetic environmental factors (multifactorial). Therefore, it is likely that sequence variation alone is not sufficient to predict the risk of disease susceptibility, particularly in homeostatic organisms like humans. Nevertheless, these variants may provide a starting point for further inquiry.

Publication Types:

- Review
- Review, Tutorial

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